

# F18-FDG-PET/CT in a patient affected by Lynch syndrome

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## Abstract

Lynch syndrome (LS) is the most common hereditary syndrome that predisposes patients to colorectal cancer, and it accounts for 2–5% of the total burden of colorectal cancer. We report a case of a 61-year-old female affected by Lynch syndrome who underwent multiple adenocarcinoma resections, studied by F18-FDG-PET/CT for 5 years. This case report suggests a potential role of F18-FDG-PET/CT in the evaluation of patients affected by Lynch syndrome.

**Key words:** PET, neurofibromatosis, malignant degeneration

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## Introduction

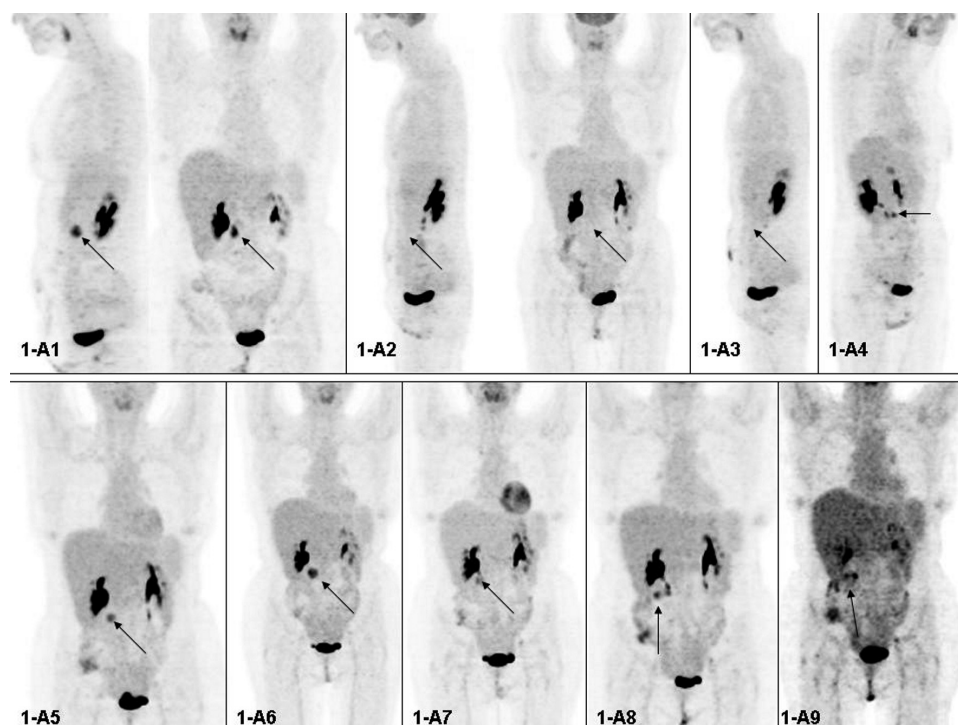
Lynch syndrome (LS) is the most common hereditary syndrome that predisposes patients to colorectal cancer, and it accounts for 2–5% of the total burden of colorectal cancer [1]. Since the identification of mismatch repair mutations in this syndrome, it has become known as Lynch syndrome and is characterized by germline mutation in a mismatch repair gene, most commonly MLH1, MSH2, or MSH6 [2]. It is now quite common to identify a germline mutation in one of the mismatch repair genes. It is characterised by an autosomal dominant inheritance pattern,

right-sided colonic cancer predilection, earlier average age of onset of colorectal cancer than in the general population, accelerated carcinogenesis, high risk of additional colorectal cancers, and increased risk of malignant disease at other sites like the pancreas, upper uroepithelial tract, endometrium, ovary, brain (Turcot's syndrome variant of LS), stomach, small bowel, hepatobiliary tract [2], and increased survival from colorectal cancer. Once a high-risk individual is recognized, management focuses primarily on strategies for risk reduction. Colonoscopic screening has been shown to significantly improve survival among carriers of a mutation associated with LS. Recommendations for at-risk members of families with LS include screening colonoscopy, endometrial sampling, transvaginal ultrasound, and in selected cases prophylactic colon and ovary or uterine surgery [3]. Surgery is the mainstay of treatment and can be prophylactic or therapeutic with curative or palliative intent. The type and extent of surgical resection is influenced by the effectiveness of, and patient compliance with, available surveillance strategies. There are no guidelines regarding the timing of surgery. Colectomy is recommended for patients with Lynch syndrome who have colorectal cancer, but the choice of procedure remains controversial as surgical options include a total proctocolectomy (TPC) with permanent ileostomy, a total proctocolectomy with ileal pouch anal anastomosis (IPAA), or a subtotal colectomy with ileorectal anastomosis (IRA) [4].

## Case report

We report the case of a 61-year-old female affected by LS studied by F18-FDG-PET/CT for 5 years. The patient underwent hysterectomy for an ovarian cancer in 1989, right hemicolectomy for adenocarcinoma in 1991, and another resection of transverse colon and part of the descending colon because of a disease relapse. In 2005 the patient developed an adenocarcinoma of the rectum and underwent extended surgical resection of the remnant colon and rectum with permanent ileostomy; a subsequent diagnosis of LS was given. In 2006 an endoscopic examination revealed an infiltrative duodenal lesion, and the patient underwent an F18-FDG-PET/CT that confirmed this finding (Figure 1-A1). A resection of the head of the pancreas and duodenum was performed and two PET studies during 2007 remained negative (Figure 1-A2; Figure 1-A3). In 2008, after a rising value of CA19.9 marker, a new

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**Figure 1.** In this figure maximum intensity projection images of all F18-FDG-PET/CTs are reported; in particular Figure 1-A1 reports left lateral and anterior views of the study performed in 2006, showing the lesion (arrows); Figure 1-A2 and Figure 1-A3 report left lateral and anterior views of the negative studies performed during 2007; Figure 1-A4 shows the site of disease in a 30° right oblique view of the study performed in 2008; Figure 1-A5 reports the anterior view of a study performed in the same year where the persistent pathologic uptake is visible; Figure 1-A6 shows the anterior view of the study performed in 2009 revealing diseases progression (arrow); Figure 1-A7 shows the anterior view of the study performed in 2009 after radiotherapy documenting an uptake reduction; Figure 1-A8 and Figure 1-A9 show anterior views of the studies performed at the end of 2009 and at the beginning of 2010 revealing disease relapse and progression.

F18-FDG-PET/CT was performed revealing pathologic uptake at a solid mesenteric lesion, just ahead of the right ureter (Figure 1-A4). Chemotherapy was started, and after two cycles another PET study revealed no significant uptake reduction (Figure 1-A5). At the beginning of 2009 another PET study documented disease progression at the end of chemotherapy at the same site (Figure 1-A6); on the basis of these findings, radiotherapy was performed with moderate effect in terms of uptake reduction in the middle of 2009 (Figure 1-A7), but, unfortunately, at the end of 2009 another PET study documented a slight increase of pathologic uptake at mesenteric lesion (Figure 1-A8) and CA19.9 elevation; at the beginning of 2010 significant disease progression in terms of higher uptake, a new mesenteric lesion near the previous one, and significant CA19.9 elevation were documented (Figure 1-A9).

## Discussion

The role of F18-FDG-PET/CT in oncologic imaging is well established; it has gained widespread acceptance as a diagnostic tool in oncology and is an increasingly important imaging test [5]. Numerous reports are available in literature about the usefulness and diagnostic role of F18-FDG-PET/CT in evaluating pancreatic, endometrial, gastro-intestinal, and ovarian malignancies [6–9]; moreover, current evidence suggests that F18-FDG-PET/CT has an established role as a routine component

of colorectal cancer (CRC) patient management in the detection of recurrent [10, 11] or residual disease and preoperative assessment prior to metastasectomy. It is likely that further evidence will support the emerging roles in the staging, radiotherapy treatment planning, and assessment of treatment response both for primary tumours and metastases [12, 13]. This evidence suggests that F18-FDG-PET/CT could be considered an imaging tool able to usefully evaluate all the organs possibly affected by malignancies in LS because of their hypermetabolic behaviour. To our knowledge there are no reports in literature about LS evaluation using F18-FDG-PET/CT, and this case report reveals how PET studies could be useful in evaluating this kind of patient both in terms of staging/restaging and therapy efficacy monitoring, possibly modifying and guiding clinical management and therapeutic decision making. In conclusion, a potential role of F18-FDG-PET/CT in the evaluation of patients affected by LS should be considered despite the fact that wider studies are needed to definitively confirm this insight.

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